

---

# COMMENTARY & PERSPECTIVE

---

## Multiple Hereditary Exostoses: Its Burden on Childhood and Beyond

Commentary on an article by A.L. Goud, MD, et al.: "Pain, Physical and Social Functioning, and Quality of Life in Individuals with Multiple Hereditary Exostoses in the Netherlands. A National Cohort Study"

Alexandre Arkader, MD

Multiple hereditary exostoses is a rare autosomal dominant condition that affects approximately one in 50,000<sup>1</sup>. The genetic mutations leading to multiple hereditary exostoses are located in the *EXT1* or *EXT2* genes, which are in part responsible for the heparan sulfate chain elongation or formation<sup>2</sup>. The type of genotypical abnormality may have direct influence in the phenotypical appearance of the affected children<sup>2</sup>. The literature is filled with studies on the etiology and genetics of this condition. There are also several reports on different surgical techniques for osteochondroma removal and for reconstruction of secondary deformities resulting from the osteochondromas. There is, however, a paucity of studies on the natural history of the disease and the quality of life of these patients as it relates to development and growth, physical activities, integration into society, and the presence of symptoms, such as pain<sup>1,3</sup>.

The study by Goud et al. aimed at evaluating the natural history and impact of multiple hereditary exostoses on the quality of life of patients as they reach adulthood. They analyzed 283 patients with multiple hereditary exostoses in the Netherlands and utilized self-assessment tools for children and adults. Among the children, approximately 60% played sports, but another almost 30% had stopped because of the multiple hereditary exostoses. The older the kids, the more likely they were to stop. Half of the children attending school had complaints, most related to physical activities but also including difficulty writing and using computers or being bullied. Approximately 60% of the children had pain (usually associated with a more negative perception of their disease), problems at school, and a greater number of surgical procedures. Among adults, approximately 50% had to stop playing sports because of the multiple hereditary exostoses. Although 65% of the adults were employed, almost 30% had to change or tailor their jobs, which included making modifications to their workplace, because of their disease. Older patients tended to have insurance-related problems, such as higher premium payments. Approximately 80% of the adults had pain. Pain was worsened by activities such as walking or lifting heavy objects, and a third of the patients needed pain medications regularly. The presence of pain related to problems at work. Almost 90% of the adults compared with less than half of the children needed at least one surgical procedure.

Darilek et al.<sup>3</sup> showed in their cross-sectional study that the prevalence of pain (diffuse, not necessarily over an exostosis) in children and adults with multiple hereditary exostoses reached approximately 80%. Approximately 80% of the patients in their study needed surgical treatment. The authors also reported a correlation between pain and the need for surgery or the presence of complications related to multiple hereditary exostoses, such as muscle or tendon impingement. Over 70% of the study cohort used some pain medication.

While multiple hereditary exostoses may be seen as a disease of the growing skeleton, it is evident that the burden of this condition extends beyond childhood and adolescence, and this burden may actually worsen throughout life. Despite the possible cultural differences between the patient population studied by Goud et al. and the patient population in North America, this study shows that there is an increase in the prevalence of pain, childbearing complications, and need for surgery throughout adulthood. These findings highlight the need for adequate counseling, long-term follow-up, and measures to improve the quality of life of patients with multiple hereditary exostoses.

Alexandre Arkader, MD\*

Children's Hospital Los Angeles,  
Los Angeles, California

\*The author received no payments or services, either directly or indirectly (i.e., via his institution), from a third party in support of any aspect of this work. Neither the author nor his institution has had any financial relationship, in the thirty-six months prior to submission of this work, with any entity in the biomedical arena that could be perceived to influence or have the potential to influence what is written in this work. Also, the author has not had any other relationships, or engaged in any other activities, that could be perceived to influence or have the potential to influence what is written in this work. The complete **Disclosures of Potential Conflicts of Interest** submitted by authors are always provided with the online version of the article.

## References

1. Schmale GA, Conrad EU 3rd, Raskind WH. The natural history of hereditary multiple exostoses. *J Bone Joint Surg Am.* 1994 Jul;76(7):986-92.
2. Jones KB. Glycobiology and the growth plate: current concepts in multiple hereditary exostoses. *J Pediatr Orthop.* 2011 Jul-Aug;31(5):577-86.
3. Darilek S, Wicklund C, Novy D, Scott A, Gambello M, Johnston D, Hecht J. Hereditary multiple exostosis and pain. *J Pediatr Orthop.* 2005 May-Jun;25(3):369-76.